KCNE2 gene

potassium voltage-gated channel subfamily E regulatory subunit 2

Normal Function

The *KCNE2* gene provides instructions for making a protein that regulates the activity of potassium channels. These channels, which transport positively charged atoms (ions) of potassium into and out of cells, play a key role in a cell's ability to generate and transmit electrical signals.

The specific function of a potassium channel depends on its protein components and its location in the body. The KCNE2 protein regulates several ion channels, including a channel made up of proteins produced by the *KCNH2* gene. Channels made with the KCNH2 protein are present in heart (cardiac) muscle, where they transport potassium ions out of cells. This form of ion transport is involved in recharging the cardiac muscle after each heartbeat to maintain a regular rhythm.

The proteins produced from the *KCNH2* and *KCNE2* genes interact to form a functional potassium channel. Four alpha subunits, each produced from the *KCNH2* gene, form the structure of each channel. One beta subunit, produced from the *KCNE2* gene, binds to the channel and regulates its activity.

Health Conditions Related to Genetic Changes

familial atrial fibrillation

A mutation in the *KCNE2* gene is associated with rare cases of an abnormal heart rhythm called familial atrial fibrillation. In a small number of people with this condition, researchers have found a mutation that replaces the amino acid arginine with the amino acid cysteine at position 27 of the protein made by the *KCNE2* gene (written as Arg27Cys or R27C). In cardiac muscle cells, this mutation appears to increase the flow of potassium ions through certain channels regulated by the KCNE2 protein. The enhanced ion transport may alter the heart's normal rhythm. Researchers are working to determine whether the R27C mutation is the direct cause of atrial fibrillation in these affected individuals.

Romano-Ward syndrome

More than 10 mutations in the *KCNE2* gene have been identified in people with Romano-Ward syndrome. These mutations change a single protein building block (amino acid) in the KCNE2 protein, which alters the protein's ability to regulate potassium channels in cardiac muscle cells. The channels open more slowly and close more rapidly than usual, decreasing the flow of potassium ions out of these

cells. This disruption in ion transport causes an abnormal heart rhythm (arrhythmia) that increases the risk of fainting (syncope) and sudden death.

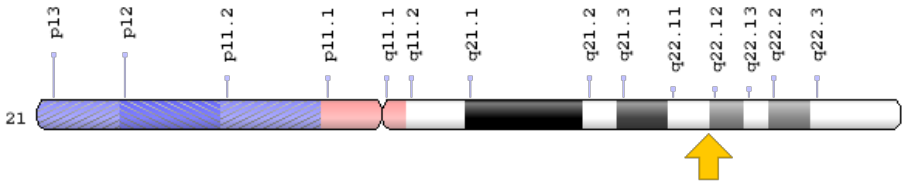
other disorders

Certain drugs, including medications used to treat arrhythmias, infections, seizures, and psychotic disorders, can lead to another type of abnormal heart rhythm in some people. This drug-induced heart condition, which is known as acquired long QT syndrome, increases the risk of cardiac arrest and sudden death. A small percentage of cases of acquired long QT syndrome occur in people who have an underlying mutation in the *KCNE2* gene.

Chromosomal Location

Cytogenetic Location: 21q22.11, which is the long (q) arm of chromosome 21 at position 22.11

Molecular Location: base pairs 34,364,024 to 34,371,141 on chromosome 21 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KCNE2_HUMAN
- LQT6
- minimum potassium ion channel-related peptide 1
- MinK-related peptide 1
- MIRP1
- Potassium channel beta subunit MiRP1
- potassium channel, voltage gated subfamily E regulatory beta subunit 2
- potassium voltage-gated channel, Isk-related family, member 2

Additional Information & Resources

Educational Resources

- Neuromuscular Disease Center, Washington University: KCNE family of potassium channels
  http://neuromuscular.wustl.edu/mother/chan.html#kcne

GeneReviews

- Long QT Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1129

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KCNE2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 2
  http://omim.org/entry/603796

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KCNE2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KCNE2%5Bgene%5D
- HGNC Gene Family: Potassium voltage-gated channel regulatory subunits
  http://www.genenames.org/cgi-bin/genefamilies/set/858
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6242
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/9992
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